OFFICE OF PURCHASING, TRAVEL AND FLEET MANAGEMENT
SOLE SOURCE DETERMINATION (SSD)

(TO BE SUBMITTED TO OPTFM UPON COMPLETION OF RFIN)

AGENCY CONTACT INFORMATION SUBMITTING SOLE SOURCE DETERMINATION

Agency Name: Jackson State University / RCMI Center for Environmental Health

Agency Contact: Dr. Paul Tchounwou Phone: 601-979-0777

Agency Contact Email Address: paul.b.tchounwou@jsums.edu

RFIN#:_____

SOLE SOURCE INFORMATION (Part A)

Vendor: Illumina

Vendor Contact: Ketan Patel

Phone: 858-202-4500

Vendor Contact Mailing Address: 5200 Illumina Way, San Diego, CA 92122

Vendor Contact Email Address: kpatel5@illumina.com

Vendor Contact Fax Number: 858-202-4766

Date Form Submitted: December 05, 2018-Commodity: Illumina NextSeq 550 System

Other Description/Information: ----------

Total Value.....$256,666.25

QUALIFYING INFORMATION (Part B)

Were any objections to the sole source determination regarding this commodity received by the Agency?

□YES X NO

If "yes", did the Agency appeal to PPRB? □YES X NO

What was the outcome of the appeal to PPRB?

'If Agency received no objections to sole source determination, complete entire form and submit with P-1 Request.

If PPRB approval received, please complete the remainder of this form and submit with P-1 Request. If PPRB did not approve, the Agency is required to submit the procurement of the commodity to an advertised competitive bid or selection process. Once competitive bid or selection process is complete, attach this form (SSD) as an internal attachment to P-1 request and do not complete (Part C).

PPRB APPROVAL RECEIVED (Part C)

What efforts were made by the Agency to determine that the proposed provider is the only entity that can provide the commodity needed?

The RCMI Center for Environmental Health at Jackson State University did a national search to identify the scientific equipment that would allow the Centers' investigators and students to perform genomics and transcriptomics research, and discovered that the NextSeq 550 System was the equipment needed for that purpose. We also found that Illumina Inc., a biotech company located in San Diego, California, USA, is the sole manufacturer and solely distributer of this analytical instrument in the United States.

What efforts were made to ensure the best possible price for the commodity was obtained?

During the grant proposal submission to the National Institutes of Health, Dr. Paul Tchounwou (principal investigator and director of RCMI Center for Environmental Health at JSU, contacted the sale representative at Illumina Inc., who provided a quote. This quote was reviewed and approved by NIH. Also, Illumina, Inc. provided a 5% academic discount toward the purchase of this equipment.

Why is the commodity the only one that can meet the needs to the Agency?

The capacities to conduct genomic-based analyses are increasingly needed to address complex research questions in environmental health sciences. We did an extensive search of the scientific literature and found that the Illumina NextSeq 550 System is the only desktop next-generation sequencing (NGS) system capable of sequencing a 30× human genome in a single run. The requested Illumina NextSeq 550 Sequencing System is an integrated state-of-the-art and high resolution next generation sequencing technology for automated generation of deoxyribonucleic acid (DNA) clonal clusters by bridge amplification, sequencing, and primary analysis. The system includes an embedded touchscreen monitor and a computer unit loaded with the NextSeq control software for system operation. It is a highly innovative next-generation sequencer that integrates amplification, sequencing and data analysis into one single instrument. By leveraging the latest advances in sequencing by synthesis (SBS) chemistry, and the industry simplest workflow, the NextSeq 550 sequencing instrument delivers high quality results for exome, transcriptome, and targeted resequencing applications. It is able to provide a rapid turnaround time and cost-effective genetic analysis for the widest range of applications. The rapid turnaround time for sequencing makes it even more attractive, as it makes possible the analysis of genetic variation and function in a very timely manner. This innovative sequencing and array technology has significantly contributed to groundbreaking advancements in life science research, and molecular diagnostics, as well as in medical, translational and public health research. In previous years investigators at the JSU RCMI Center for Environmental Health who needed to perform RNA sequencing were required to initiate their experiments at JSU, extract RNA samples and then ship them out for RNA sequencing analysis at commercial laboratories. With the acquisition of this high-end instrument, RCMI investigators at JSU will be able to address biomedical and health science issues through pioneering research and education in genetics, genomics and transcriptomics; and to educate and train a new generation of underrepresented minorities in innovative and translational applications of genetics science to medicine, public health and society.

Why is the amount expended for the commodity reasonable?

The budgeted amount to purchase the NextSeq 550 Sequencing System is reasonable because the researchers at JSU RCMI Center for Environmental Health spend an average of $50,000 per year to pay commercial laboratories for the analysis of the RNA samples. Hence it is more economical to have the System available in-house to facility research implementation, improve efficiency and reduce costs. The critical need for this instrument has been reviewed and approved by the National Institutes of Health that provided the funds for its purchase. Having the instrument available on the JSU campus will not only promote the development and implementation of genomics/biomedical research but also facilitate the research training of the next generation of biomedical scientists.

<u>The following items must be attached to your P-1:</u>

1. Letter from Agency head, or designee, outlining the results of the procedures that have been detailed above.
2. Copy of SSD form and any accompanying documents.
3. Copy(s) of OSS, if applicable.
4. Original quote, signed by Vendor. Electronic quotes do not require the signature of the Vendor.

   See 31-7-13(b) or 3.108.03.1 of Procurement Manual.

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Thursday, November 15, 2018

RCMI-Center for Environmental Heath
Jackson State University
JSU Box. 17131
Jackson, Mississippi 39217-0940

Dear RCMI-Center for Environmental Heath,

Re: Justification Sole Source Letter: NextSeq 550 System

The NextSeq 550 System from Illumina is the only commercially available system of its type. It is a mid-throughput, integrated desktop platform that uses massively parallel sequencing technology and arrays for genetic analysis and functional genomics. NextSeq 550 technology utilizes prepared libraries which are loaded directly on the system. Integrated cluster generation provides automated clonal amplification of single molecules randomly distributed on a glass surface. Resulting DNA clusters are sequenced on the NextSeq 550 System using the Illumina sequencing by synthesis (SBS) method with patented reversible terminator chemistry. The system also includes an option to integrate with the BaseSpace® genomics computing environment, an easy, secure, and cost-effective way to store, analyze, and share genomic data, available as either a cloud or an onsite deployment (BaseSpace Onsite system).

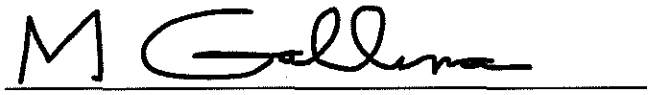
The Illumina NextSeq 550 System is the only desktop next-generation sequencing (NGS) system capable of sequencing a 30× human genome in a single run. Additionally, the NextSeq 550 System is the only desktop system capable of both NGS and microarray scanning, transitioning seamlessly between the 2 technologies.

The NextSeq 550 System enables 20–120 Gb of data output in a single run, providing flexibility across the broadest range of genomic applications and study sizes. Its simple workflow and quick run times provide fast desktop sequencing of exomes, transcriptomes, and whole genomes. In addition to its sequencing capabilities, this desktop platform provides Infinium® HumanCytoSNP- 12, CytoSNP-850K, and HumanKaryomap-12 BeadChip scanning on the same instrument.

Illumina has numerous patents, copyrights, and other intellectual property rights with respect to the Illumina products.

The NextSeq 550 System is available only from Illumina and its authorized distributors.

Sincerely,



MariJo Gallina
Staff Product Manager, NextSeq
Illumina, Inc.,

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QUOTATION FOR SUPPLY OF GENETIC ANALYSIS PRODUCTS

Prepared by:

Illumina, Inc.
5200 Illumina Way
San Diego CA 92122-4616
USA
Hereinafter referred to as "Illumina"

Prepared for:

Paul Tchounwou

Jackson State University

Hereinafter referred to as " Jackson State University " or "Customer"

Quotation Number:	**4169556**
Quotation Date:	**Oct 22, 2018**
Expiration Date:	**Nov 21, 2018**
Prepared By:	**Ketan Patel**
Phone Number:	**+1 (858) 754.4493**
Email:	**KPATEL5@ILLUMINA.COM**

QUOTATION NUMBER	4169556	**PREPARED BY**	Ketan Patel
QUOTATION DATE	Oct 22, 2018	**TEL**	+1 (858) 754.4493
EXPIRATION DATE	Nov 21, 2018	**EMAIL**	KPATEL5@ILLUMINA.COM
CURRENCY	USD		

I. CUSTOMER INFORMATION

Company or Institution Name:	Jackson State University
Customer Number:	6000025974
Address:	Jackson State University 1400 John R. Lynch St Jackson MS 39217-0940 USA
Contact Name:	Paul Tchounwou
Phone:	(601) 979-0777
E-Mail:	paul.b.tchounwou@jsums.edu
Ultimate Consignee:	Jackson State University
Bill to address	Jackson State University 1400 John R. Lynch St Jackson MS 39217-0940 USA
Shipping address:	Jackson State University 1400 John R. Lynch St Jackson MS 39217-0940 USA

II. PRODUCT & PRICING INFORMATION

Catalog#	Product Description	Unit Price (USD)	Discount %	Customer Price (USD)	Units	Subtotal (USD)
SY-415-1002	NextSeq® 550 Sequencing System: Illumina NextSeq 550 Sequencing System is an integrated system for automated generation of DNA clonal clusters by bridge amplification, sequencing, primary analysis, and array scanning. System includes embedded touchscreen monitor and on-instrument computer, NextSeq Control Software, installation and training, and 12 months warranty (including parts and labor).	275,000.00	9.00	250,250.00	1	250,250.00
Subtotal						250,250.00
Freight / Handling Fee						2,502.50
Total Fees (including shipping and insurance)						**252,752.50**

Notes: Tax is an estimate and is subject to change upon invoicing based upon the appropriate tax regulations.

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QUOTATION NUMBER	4169556	**PREPARED BY**	Ketan Patel
QUOTATION DATE	Oct 22, 2018	**TEL**	+1 (858) 754.4493
EXPIRATION DATE	Nov 21, 2018	**EMAIL**	KPATEL5@ILLUMINA.COM
CURRENCY	USD		

III. CONDITIONS OF SALE

By submitting an order, Customer accepts and agrees that the Terms and Conditions referenced in this Quotation(as set forth in the section VII herein (Terms and Conditions) is the sole and exclusive agreement between Customer and Illumina with respect to the Illumina products and/or services as described above and accepts all other terms of this quotation. NOTWITHSTANDING THE FOREGOING, IF ILLUMINA AND CUSTOMER HAVE ENTERED INTO A VALID AND ENFORCEABLE AGREEMENT GOVERNING THE ILLUMINA PRODUCTS AND/OR SERVICES DESCRIBED ABOVE, THE ORDER OF PRECEDENCE BETWEEN THE AGREEMENT AND THE TERMS AND CONDITIONS SHALL BE AS FOLLOWS: IN THE EVENT OF A CONFLICT BETWEEN THE TERMS OF THE AGREEMENT AND THE TERMS AND CONDITIONS, OR IF THE AGREEMENT INCLUDES ADDITIONAL TERMS NOT ADDRESSED IN THE TERMS AND CONDITIONS, THE AGREEMENT SHALL GOVERN WITH RESPECT TO SUCH TERMS. Illumina does not supply plastics such as microplates or pipette tips for use in the listed assays and these are not included in the consumables pricing provided; however, as a result of the highly multiplexed nature of all assays, plastics alone contribute minimally to the final cost.

Customer and Illumina agree as follows:
- Customer's purchase of the products referenced in this Quotation is not conditioned on future performance characteristics or applications, whether or not realized.
- Unless otherwise agreed by Illumina in writing, Illumina will not assist Customer in developing, testing, or validating unsupported applications.
- Illumina will not replace any consumables or reagent kits if the cause of any performance failure is due to unsupported applications.
- Illumina is unable to provide any assurances or guarantee that the performance of the products referenced in this Quotation will match published specifications when used for unsupported applications.

IV. SHIP HOLD

In cases where this Quotation does not include a pre-defined ship schedule, the following ship hold terms shall apply:
- All orders must have a defined ship schedule. The initial ship date must be no later than three (3) months from the date the purchase order is received by Illumina (as provided in the Order Confirmation) and the entire order must be shipped complete within twelve (12) months from Illumina's receipt of the purchase order.
- Any exceptions to these ship hold terms must be agreed to in writing by Illumina and the Customer must pre-pay at least fifty percent (50%) of the purchase order amount of the affected shipments.
- Customers may request two (2) shipment delays for any single purchase order. The total months of delayed shipment for shipments associated with a single purchase order shall not exceed six (6) months.
- If Customer has requested a delayed shipment, Illumina reserves the right to change the lead time necessary to initiate Customer's first shipment (which may be longer than the lead time quoted at the time of the order placement).
- If Customer cannot take shipment in accordance with these terms, Illumina reserves the right to cancel the order in its entirety without any liability to the Customer.

V. HOW TO ORDER

For all consumable orders: Please submit your order online through MyIllumina (http://my.illumina.com).	For all other orders: Please submit your institutional Purchase Order and a complete copy of this quotation to the attention of: Illumina Customer Service customerservice@illumina.com Phone: +1.858.202.4566 Toll Free: +1.800.809.ILMN (4566) Fax: +1.858.202.4766

Order Confirmation

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You will receive an e-mail confirmation containing your order number within 1 business day. Another email will be sent to notify you when your order has been shipped.

VI. EXPIRATION OF OFFER

The offer contained in this document is revocable at the sole discretion of Illumina if not executed by Customer and a purchase order received by Illumina before 5:00 pm Pacific Time on the expiration date shown on page 1 of this quotation.

VII. TERMS AND CONDITIONS

By this Quotation, Illumina conditionally offers to Customer the Illumina products and/or services as described above. This offer is conditional on, and may only be accepted by, Customer's agreement that Illumina's terms and conditions listed below or otherwise included with the product or service, as applicable to the specific product or service quoted ("Terms and Conditions"), is the sole and exclusive agreement between Customer and Illumina with respect to the particular products or service. For the avoidance of doubt, any terms and conditions applicable to "Test Specific Products" that have a specific intended use in such products' documentation, as set forth in such terms and conditions, are applicable only to in vitro diagnostic products.

http://www.illumina.com/content/dam/illumina-marketing/documents/terms-conditions/united-states/usa-terms-and-conditions-of-sale-general.pdf.

Additionally, if Customer is purchasing Illumina professional consultingservices as relate to instruments, Customer environment or workflows (inall cases, excluding instrument warranty services) ("ProfessionalServices"), Customer agrees such Professional Services are exclusivelygoverned by the Terms and Conditions - Services (Professional Services)located here:

http://www.illumina.com/content/dam/illumina-marketing/documents/company/terms-and-conditions-services.pdf

In the case of BaseSpace Clarity LIMS, the Terms and Conditions are https://www.illumina.com/content/dam/illumina-marketing/documents/terms-conditions/worldwide/lims/on-premise-subscription-agreement.pdf (for subscription software), and https://www.illumina.com/content/dam/illumina-marketing/documents/terms-conditions/worldwide/lims/perpetual-license-agreement.pdf (for perpetual license software).

Additionally, notwithstanding the Illumina entity that is listed on the cover page of this Quotation, the parties understand and agree that in the event another Illumina affiliate provides the products or services to Customer, such Illumina affiliate is the relevant quoting and contracting entity under this Quotation and the relevant Terms and Conditions.

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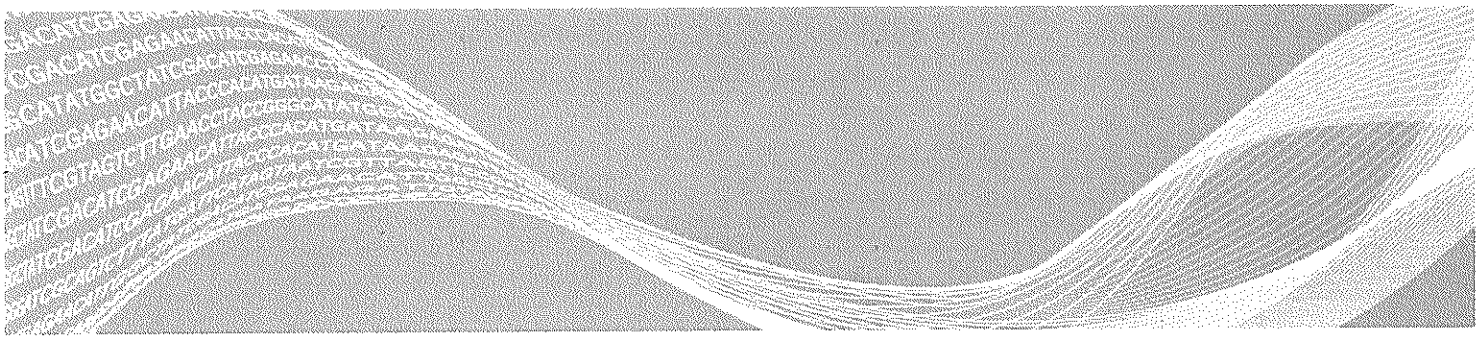
NextSeq 550

System Guide



Document # 15069765 v04

May 2018

For Research Use Only. Not for use in diagnostic procedures.

NextSeq® 550 System
Sole Source Specifications

The Illumina NextSeq 550 System is the only desktop next-generation sequencing (NGS) system capable of sequencing a 30× human genome in a single run. Two flow cell formats and multiple reagent configurations enable 20–120 Gb of data output in a single run, providing flexibility across the broadest range of applications and study sizes. Its simple workflow and quick run times enable fast desktop sequencing of exomes, transcriptomes, and whole genomes.

The NextSeq 550 System is an integrated platform that uses massively parallel sequencing technology for genetic analysis and functional genomics. Prepared libraries are loaded directly on the system. Integrated cluster generation provides automated clonal amplification of single molecules randomly distributed on a glass surface. Resulting DNA clusters are sequenced on the NextSeq 550 System using the Illumina sequencing by synthesis (SBS) method with patented reversible terminator chemistry.

The system consists of the NextSeq 550 sequencer, which incorporates a suite of dedicated flow cell and reagent configurations to generate up to 400 million clusters passing filter (up to 120 Gb) in the High Output configuration and up to 130 million clusters passing filter (up to 40 Gb) in the Mid Output configuration. The system also includes an option to integrate with the BaseSpace® genomics computing environment, an easy, secure, and cost-effective way to store, analyze, and share genomic data, available as either a cloud or an onsite deployment (BaseSpace Onsite system).

In addition, the NextSeq 550 System is the only desktop system capable of both sequencing and BeadChip array scanning, transitioning seamlessly between the 2 technologies. For scanning, prepared BeadChips are loaded onto the array adapter and placed in the same position as a flow cell. After scanning, an image file is generated for off-instrument analysis. Currently, the NextSeq 550 System supports Infinium® HumanCytoSNP-12, CytoSNP-850K, and HumanKaryomap-12 BeadChips.

The NextSeq 550 System offers:

- Scalability (20–120 Gb) in a single run to support a broad range of applications and study sizes
- Sequencing runs, including on-board cluster generation, complete in 12–30 hours
- Fully automated on-board cluster generation enables prepped libraries to be loaded directly onto the instrument
- High accuracy using Illumina SBS
- Proven SBS chemistry with single-base extension enables accurate sequencing of homopolymers
- Fully automated paired-end sequencing
- Automated BeadChip array scanning and image file generation
- Approximately 7000 peer-reviewed publications have been published using Illumina SBS sequence data, and more than 24,000 peer-reviewed publications have been published using Illumina array technology

Table of Contents

System Workflow and Applications

Feature	Description
Easy, simple, and integrated workflow	**Single, integrated instrument for sequencing and array scanning** • Single instrument performs clonal amplification, sequencing, paired-end turn, and primary data analysis (eg, base calling) • Scanning of BeadChip arrays produces image file for data analysis • Alignment, variant calling, and reporting are supported in BaseSpace, the Illumina genomics computing environment • Instrument footprint requires less than 2 square feet of benchtop space • No need for dedicated ancillary amplification system or computing/IT infrastructure • Does not require emulsion PCR
Short hands-on time	**12-hour total sequencing time with less than 10 minutes total hands-on time** • 10 minutes hands-on time for run set up (amplification, paired-end sequencing, or array scanning) • 12-hour sequencing time (1 × 75 bp) including on-instrument cluster generation, sequencing run, and automated post-run wash
Paired-end capability	• Hands-free, automated, on-instrument paired-end sequencing • Supports up to 2 × 150 bp read length
High output	• Up to 120 Gb of high-quality data passing filter per 2 × 150 bp run • Only desktop instrument capable of sequencing a 30× human genome in a single run
Flexible reagent configurations	• Multiple flow cell and reagent configurations enable sequencing of 20–120 Gb per run, providing the highest sample size and application flexibility of any desktop sequencer • Up to 400 M single read clusters (800 M paired-end reads) passing filter in the High Output configuration enable up to 120 Gb per run • Up to 130 M single-read clusters (260 M paired-end reads) passing filter in the Mid Output configuration enable up to 40 Gb per run
Most accurate data quality	**Accurate variant detection enabled by proven SBS chemistry** • Competitive nucleotide addition with a proprietary reversible terminator technology allows for highly accurate sequencing, even through homopolymeric regions. SBS chemistry is the demonstrated leader in data accuracy. [1–5] **Exceptionally high quality score distributions** • > 75% of bases with Q-scores > 30 (2 x 150 bp)

Feature	Description
Wide application flexibility	**Widest application flexibility among all desktop sequencers** • Up to 120 Gb of output allows sequencing of a 30× human genome • Up to 800 M paired-end reads enables multiplexed sequencing of exomes and transcriptomes in a single run • Up to 400 M single reads enables gene expression profiling and cytogenetic applications • Variable length amplicon and enrichment panels spanning hundreds to thousands of base pairs • Cost-effective, flexible options for sequencing runs with smaller number of samples using Mid Output kits • Additional array scanning functionality • Approximately 7000 peer-reviewed publications using reversible terminator-based SBS chemistry • More than 24,000 peer-reviewed publications using BeadArray™ technology

Instrumentation and Software

Feature	Description
Data generation	**Number of reads per run** • Up to 400 million reads per run (eg, clusters passing filter) using High Output flow cell • Up to 130 million reads per run (eg, clusters passing filter) using Mid Output flow cell **Throughput per run** • 100–120 Gb data per 2 × 150 bp run using High Output flow cell and reagents • 33–40 Gb data per 2 × 150 bp run using Mid Output flow cell and reagents **Sequencing flexibility** • Set-up options include single-read or paired-end runs • Flow cell options (eg, Mid or High Output flow cells) can be used to select data output levels • Read length is fully adjustable up to 300 base pairs **Array scanning** • Single array imaged per scanning session • BeadChip scanning in 40 minutes • Up to 12 samples imaged per run with current array support

Feature	Description
Instrumentation	**NextSeq 550 System physical specifications** • Desk-top instrument; W×D×H: 58.5 cm × 53.4 cm × 63.5 cm (23.0 in × 21.0 in × 25 in) • Weight: 83 kg (183 lbs) **Illumination** • 12 light-emitting diodes at 520 nm, 650 nm **Reagent handling** • Reagent compartment has capacity for 1 reagent cartridge containing reagents for cluster generation, paired-end chemistry, and up to 300 cycles of sequencing • Reagents arrive premixed in an integrated, RFID enabled, reagent cartridge **Sample loading** • Libraries are loaded directly into the sequencer through an on-board template loading station **Flow cells** • NextSeq 550 System is a single flow cell system • Each flow cell is a substrate with a single channel that can be imaged on both the top and bottom surfaces **Flow cell loading** • Flow cells are auto-positioned • Flow cells are keyed such that there is only 1 correct orientation **Array adapter loading** • BeadChips are placed into the array adapter • Array adapter is placed on the stage for BeadChip scanning **Instrument control computer** • Instrument control computer is integrated in the sequencer—no additional computer purchase is required • Dual Intel Xeon E5-2448L 1.8 GHz CPU with 96 GB of RAM included for instrument control, processing images, and base calling • Conducts real-time analysis processing that automatically produces image intensities and quality-scored base calls directly on the instrument computer • Sequence output contains accurate base calls and qualities derived directly from intensity data and not from a reference, sequence-based, or multiple-color encoding scheme

Feature	Description
Sequencing run time	**Sequencing runs can be completed in*:** • ~ 12 hours for a 1 × 75 bp single-read sequencing run • ~ 18 hours for a 2 × 75 bp paired-read sequencing run • ~ 30 hours for a 2 × 150 bp paired-read sequencing run *Times include cluster generation, sequencing with High Output flow cell, and base calling with quality scores.
Daily throughput	Up to 120 Gb of high-quality filtered bases in 29 hours on the sequencer (2 × 150 bp reads) or up to ~100 Gb per day.
Instrument control software	**NextSeq Control Software (NCS) offers a simple interface to configure, launch, and monitor sequence runs and array scanning.** • Easy-to-use, intuitive interface of NCS requires minimal training to configure, launch, and monitor runs • NCS includes Real-Time Analysis (RTA) software that automatically produces image intensities and quality-scored base calls directly on the NextSeq 550 System • NCS includes software to scan arrays and process intensities, generating an image file for downstream analysis • RTA provides the smallest data footprint with an option to compress base calls and quality scores more than 50% without loss in accuracy or variant calling performance [eg, a compressed 30× genome build is 48 GB (Gigabytes) compared to 110 GB uncompressed] • Quality statistics from 1 or multiple runs can be monitored in real time from any location using SAV (Sequencing Analysis Viewer). The SAV software is designed for Windows-based PCs, on or off the sequencing instrument • Multiple, standardized data formats ensure compatibility with downstream analysis and visualization tools • As an option, the BaseSpace genomic computing environment can be used to manage the NextSeq 550 System sequencing workflow (samples, experiments, runs, analysis) and store NextSeq 550 System data in a secure and cost-effective way, in the cloud or on premises

Feature	Description

Analysis software	• Real-Time Analysis (RTA) provides real time, on-instrument image processing, with base calling BaseSpace Apps providing analysis, including alignment and variant calling
	• Fully optimized analysis solutions within BaseSpace, which outputs easy-to-read reports for:
	  o Whole-genome resequencing
	  o Nextera® Rapid Capture Exome
	  o Transcriptome sequencing
	  o Somatic variant detection
	• Transcriptome analysis with the widely used TopHat/Cufflinks suite of tools
	• Alignment and variant calling using industry standard BWA/GATK or the Illumina Isaac™ pipeline
	• No bioinformatics skills needed to generate SNPs and indels
	• Produces FASTQ, BAM, VCF, and txt formatted files for maximum compatibility with third-party downstream software packages
	• Produces array image file, which is then imported into BlueFuse® Multi Software

Sequencing Chemistry (Illumina SBS Chemistry)

Feature	Description
Most successful and widely adopted sequencing chemistry worldwide	**Powered by TruSeq®, sequencing by synthesis chemistry** • Uses reversible terminators and a highly efficient DNA polymerase • The DNA polymerase is modified for efficient addition of nucleotides with cleavable fluorescent dyes and reversible terminators • Sequencing reactions are performed on the surfaces of a multichannel flow cell • Fluorescent dyes on the nucleotides are cleaved after imaging • Reversible terminators are removed to allow chain extension • Competitive addition from a pool of all 4 reversible terminator nucleotides • Sequenced DNA templates are copied to generate complementary strands, enabling paired-end sequencing

Library Preparation

Feature	Description

NGS library preparation kits and BeadChip arrays	**Ready-to-use kits are available to prepare libraries for sequencing or arrays:**
	• DNA sequencing (single-read, paired-end, or mate-pair reads)
	• RNA sequencing [stranded total RNA, stranded mRNA, and small RNA (microRNA)]
	• Targeted sequencing (human exome, cancer panels, custom enrichment, and custom amplicon)
	• ChIP-sequencing
	• Sample multiplexing
	  o Up to 24 indexes with TruSeq® DNA Library Prep Kits (TruSeq Nano, TruSeq PCR-Free)
	  o For highly multiplexed amplicon sequencing—up to 1536 targets per reaction, 24 samples per run with TruSeq Custom Amplicon
	• Infinium HumanCytoSNP-12 BeadChip
	• Infinium CytoSNP-850K BeadChip
	• Infinium HumanKaryomap-12 BeadChip
	Plus additional applications as developed.

Paired-end read support	Automated paired-end support for 200–350 bp insert sizes
Low sample input	• 1 µg for TruSeq DNA PCR-Free samples • 100–200 ng DNA for TruSeq Nano DNA • 50 ng for Nextera Rapid Capture Exome • 5 ng DNA for TruSeq ChIP-Seq applications • 100 ng of total RNA for TruSeq RNA v2, TruSeq Stranded mRNA, and TruSeq Stranded Total RNA • 5 ng of total RNA for TruSeq RNA Access • 1 µg total RNA for TruSeq small RNA sequencing • 10 pg of total RNA with SMARTer Ultra Low RNA Kit • 50 ng DNA for whole-genome bisulfite sequencing

Amplification

Feature	Description
Amplification method	• Solid-phase isothermal amplification to produce clonal, single-molecule array clusters is automated, requiring no user intervention • No need for emulsion PCR or additional equipment—amplification is performed directly on the instrument
Amplification sample throughput	• TruSeq sample libraries can be prepared in a single 8-hour day or less by 1 full-time employee (FTE) for short-insert paired-end runs.
Amplification time	• A single operator can amplify up to 24 samples on a single channel in ~2 hours using a single NextSeq 550 instrument • Amplification is fully automated, using an integrated reagent cartridge with premixed and prefilled reagents
Cluster generation	• Automated simultaneous clonal amplification of hundreds of millions of single molecule DNA templates, producing clusters containing approximately 500–1000 identical copies of each original DNA sequence

Applications

Feature	Description

The most widely published sequencing chemistry with approximately 7000 peer-reviewed publications	• Whole-genome resequencing
	• Targeted resequencing including, but not limited to the following methods:
	  ○ Exomes (37 Mb–62 Mb)
	  ○ Custom enrichment panels (100s kb–Mb's)
	  ○ Custom amplicon panels (100s kb)
	• *De novo* sequencing
	• Mate pair sequencing for libraries with 2–5 kb insert sizes
	• ChIP-Seq of sequence-specific DNA binding proteins
	• ChIP-Seq of histone modifications and epigenetic marks
	• Sequencing of bisulfite-treated DNA to study DNA methylation
	• mRNA sequencing
	• Tag-based gene expression
	• Small RNA sequencing
	• Total RNA sequencing (coding + noncoding)
	• Targeted RNA sequencing
	• Ribosome profiling
	• HLA sequence-based typing
	• DNase 1 hypersensitivity site mapping
	• Nucleosome positioning and chromatin structure studies
	• ChIP-Seq: studying sequence-specific protein-RNA interactions
	• CNV-Seq: measuring copy number variation (CNV) with sequencing
	• GRO-Seq: studying RNA polymerase initiation events
	• Paired-end mRNA sequencing to study gene fusions in cancer
	• Prenatal screening from maternal blood
	• Sequencing of ancient DNA samples
	• DNA imprinting and allele-specific expression
	• Plus additional applications as developed

Established microarray technology with over 24,000 peer-reviewed publications	• Copy number variation and loss of heterozygosity (LOH) events

Informatics

Feature	Description
Automated bioinformatics delivers biologist-friendly data analysis	• Graphical user interface (no command-line interface) • No bioinformatics skills needed to generate biologically relevant data • Simple, easy-to-read reports automatically delivered • Easily import NextSeq 550 System output files to your favorite third-party software for downstream analysis • Offline data analysis is available in BaseSpace cloud or on-site environments • Reports viewable in Windows, Mac, or Linux environment • No separate computer hardware needed • Automated transcriptome analysis with the widely used TopHat/Cufflinks suite of tools • Automated DNA alignment and variant calling using industry-standard BWA/GATK or the Illumina Isaac pipeline
Seamless integration with BaseSpace, the Illumina genomic computing platform	• BaseSpace is a software with an easy to use single user interface to manage the end-to-end, sample-to-results NextSeq workflow • BaseSpace is available as both a cloud and an on-premises package (BaseSpace Cloud and BaseSpace Onsite, respectively) • BaseSpace can be accessed with a web browser from any location with access to the BaseSpace environment • Samples, experiments, and NextSeq 550 System runs can be prepared and managed in a few clicks, and algorithms checking for index compatibility and syntax errors minimize risk • Illumina provides technical support for the entire NGS workflow, from sample to answer • Remotely monitor your NextSeq 550 System run on the internet in real time via BaseSpace • Minimal bioinformatics expertise needed to perform sophisticated bioinformatics analysis • Data upload to BaseSpace is completed at end of run, eliminating time-consuming manual data transfers • Automatic data analysis (mapping, alignment, variant calling) in BaseSpace and can be set up with minimal training and without the use of command line • Whole-genome and exome sequencing using either the industry-standard BWA/GATK method, or the fast and accurate Illumina Isaac pipeline • Somatic variant detection using a tumor-normal whole-genome based combined calling analysis • Comprehensive transcriptome analysis including gene expression profiling, mRNA-Seq, and total RNA-Seq (transcript-level gene expression, detection of gene fusions, novel isoforms, cSNPs) using the industry-standard TopHat/Cufflinks workflow • With data in BaseSpace, users can instantly share their NGS data as soon as it is generated—no manual and time-consuming transfer of files is required to share run QC information with Illumina tech support or colleagues or to share sequencing data between collaborators

Feature	Description
BaseSpace Cloud	• No upfront computer hardware/infrastructure investment • Scalable data storage and archiving; limitless storage space available • Secured AES-256 encrypted data streaming and storage • EU Safe Harbor certified to facilitated data sharing with EU collaborators • Access to BaseSpace Apps featuring bioinformatics applications developed by the academic and commercial community • Works with consumer-grade internet bandwidth (~1.5 Mbps) • Instantly share data with your collaborator across the hall or across the globe with a few clicks • High availability and uptime surpassing many institutional service level agreements
BaseSpace Onsite	• BaseSpace Onsite is a server on which a local version of BaseSpace is deployed to allow NextSeq 550 System users to benefit from many of the BaseSpace features available in the cloud while keeping all their data on premises • No connection to the public internet is required • An option to encrypt the data at rest with an AES 256-bit key is provided • Data between nodes is secured on a private network • Up to 6 servers can be connected to each other to increase processing power and storage while keeping a single software environment • A storage system with redundant high performance disk array (RAID 5) for data archival can be purchased from Illumina as an accessory • Any Network Attached Storage can be used for data archival • The BaseSpace Onsite server is available for purchase with or without a 20U cabinet • OS and data drives are redundant and hot swappable, virtually eliminating the risk of downtime in the event of drive failure • Power supplies and cooling fans are redundant, virtually eliminating the risk of downtime in the event of failure • Configuration and BIOS are under strict revision level control to minimize to facilitate support by minimizing the variation in configuration between systems • Illumina staff provide installation and training
BlueFuse Multi Software	• Supplies laboratories with a single software solution for preimplantation genetic screening (PGS) and preimplantation genetic diagnosis (PGD) • Securely store and retrieve sample information and associated reports • Analyze array-based molecular cytogenetic and *in vitro* fertilization (IVF) data • Multi-user software for analyzing, visualizing, and interpreting results from molecular cytogenetics studies
GenomeStudio® Software	• Estimate LogR ratio and B allele frequency for copy number analysis • Analyze CNV data across markers • Generate a chromosomal heat map for examining copy number aberrations across the entire genome for multiple samples

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